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RHODE ISLAND DEPARTMENT OF HEALTH



RHODE ISLAND BIRTH DEFECTS PROGRAM

BIRTH DEFECTS DATA BOOK

RHODE ISLAND DEPARTMENT OF HEALTH

## ACKNOWLEDGEMENTS

RHODE ISLAND BIRTH DEFECTS ADVISORY COUNCIL  
RHODE ISLAND CHAPTER, MARCH OF DIMES  
WOMEN AND INFANTS HOSPITAL  
RHODE ISLAND HOSPITAL  
HASBRO CHILDREN'S HOSPITAL  
RHODE ISLAND KIDS COUNT  
RHODE ISLAND PARENT INFORMATION NETWORK (RIPIN)  
VNA OF CARE NEW ENGLAND



*Dear Colleague:*

*The Department of Health works to ensure that all Rhode Islanders can live safe and healthy lives. The Division of Family Health focuses on the health and development of Rhode Island's children and families. Over many years, we have helped build family-centered resources and services that recognize and respond to children's needs in their homes, medical practice, child care, schools, and other settings. Good information on the health of children and families is a cornerstone of our mission. Therefore, this report on birth defects in Rhode Island is an especially important resource.*

*The Department of Health is mandated to describe the occurrence of birth defects in children, detect trends of morbidity and mortality, and identify children with birth defects to intervene on a timely basis for treatment. Specifically, the Division of Family Health is mandated to provide and promote family-centered, coordinated care for these children and to facilitate the development of community-based systems of service for them and their families. These values are key to ensuring medical homes for Rhode Island's most at-risk children, youth, and their families.*

*These data will help focus our work and investments on the issues that are important for families raising children with birth defects. Among the Birth Defects Program's strongest priorities are:*

- » Understanding the overall prevalence and demographics of children with birth defects in our state.*
- » Assuring that children and their families receive appropriate services and referrals.*
- » Identifying and closing gaps in services and systems for families of children with birth defects.*

*We hope this information will raise awareness of the need for medical homes for children with birth defects and lead to policy changes to ensure they receive the care they need. Please join us in this endeavor to move these data to action.*



## WHAT ARE BIRTH DEFECTS?

Birth defects are structural abnormalities that affect the development of organs and tissues of an infant or child. These abnormalities may be identified during pregnancy, at birth, or following birth. Possible causes or contributing factors to birth defects include genetic (inherited) factors, environmental pollutants, occupational hazards, dietary factors, medications, and personal behaviors.

## WHY STUDY BIRTH DEFECTS?

Each year in the United States, 120,000 babies are born with a birth defect and of these, 8,000 (6.7%) die during the first year of life. Many babies who do survive experience childhood illness and disability.

Early recognition and response to birth defects often prevents more serious effects. An active birth defects surveillance and information system is essential for the development of programs and policies that can reduce birth defects and infant mortality.

Approximately 20% of all infant deaths in the United States are due to birth defects. Compared to the United States, a slightly higher proportion of Rhode Island infants (nearly one in four) die as a result of a birth defect. Of the 91 infants who died in Rhode Island during 2002, 22 of the deaths (24.2%) were attributed to a birth defect. During 1998-2002, the proportion of infant deaths resulting from a birth defect and the birth defects specific infant mortality rate have decreased. Specifically, in 1998, 24 of the 88 deaths among Rhode Island infants (27.3%) resulted from a birth defect, yielding a birth defects specific infant mortality rate of 190.5 per 100,000 live births. Since then, the Rhode Island birth defects specific infant mortality rate decreased by 10.6% to 170.3 in 2002.

Many infants who are born preterm (prior to 37 weeks gestation) also have birth defects. Research is currently being conducted nationally to determine the association of preterm births and birth defects. In Rhode Island, 158 (10.9%) of the 1,454 babies born preterm in 2002 had a birth defect. Preterm births are also the leading cause of infant deaths in Rhode Island. Of the 91 infant deaths in 2002, 25 (27.5%) were a result of prematurity. Although not the cause of death, 65 (71.4%) of the infants who died in Rhode Island were born preterm, and 56 (61.5%) were born weighing less than 1,500 grams (3lbs 5oz).

## RHODE ISLAND BIRTH DEFECTS PROGRAM: A PROGRESS REPORT

Rhode Island began developing a birth defects surveillance system in 2000, with funding from the Centers for Disease Control and Prevention (CDC). The Rhode Island Birth Defects Program is located at the Rhode Island Department of Health, Division of Family Health. The program was created to identify newborns with birth defects; to assure that these children receive appropriate preventive, specialty, and other health care services; and to monitor trends.

During 2003, the Rhode Island General Assembly enacted legislation (General Laws 23-13.3) requiring the development and implementation of a birth defects reporting, surveillance, and information system. This system will describe the occurrence of birth defects in children up to age five; detect trends of morbidity and mortality; and help assure that children with birth defects receive services and treatment on a timely basis.

The Rhode Island Birth Defects Advisory Council was appointed by the Director of Health to advise the Department on the establishment and implementation of the system and recommend a list of birth defects to be reported to the surveillance system. It is critical that state agencies, health care services providers, community organizations, parents and other key stakeholders provide input not only for the development of the surveillance system, but also for issues regarding information dissemination and analyses.



## CASE ASCERTAINMENT AND DATA

The Rhode Island Birth Defects Program has been using hospital discharge data to identify babies born with birth defects because it is the only data set that captures diagnoses coded by the International Classification of Diseases (ICD) system. This coding system, which is in its 9th Clinical Modification (ICD 9-CM), provides more specificity on the type of birth defect. Although the birth certificate contains some information on birth defects, it does not capture all of them and does not include ICD codes. A higher number of babies with birth defects have been identified using the hospital discharge database than the birth certificate. For example, during 2000, 781 babies were identified with birth defects using hospital discharge data compared with 372 using birth certificate data.

In addition to collecting data through the hospital discharge database, the Rhode Island Birth Defects Program is also working with Women and Infants Hospital, Rhode Island Hospital, and Hasbro Children's Hospital to obtain additional cases of birth defects and information on services provided to families of children with birth defects.

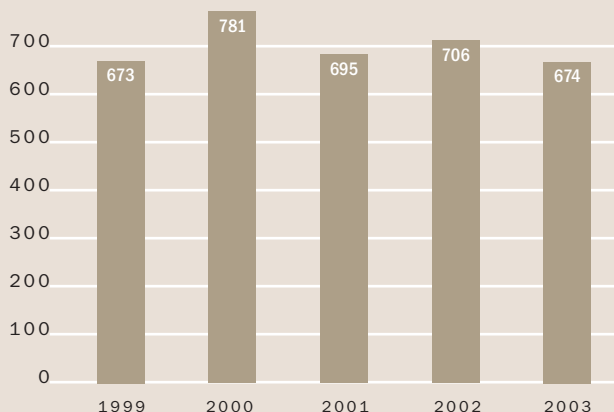
Hospital discharge data indicate that during 1999-2003, 3,529 (5.7%) of the 61,420 babies born to Rhode Island residents in Rhode Island maternity hospitals had at least one birth defect.

Figures 1 and 2 show that overall, the number and rate of birth defects in Rhode Island have remained fairly stable over the past five years. Of Rhode Island's 13,000 annual births, approximately 700 babies are born with birth defects. This is an average birth defect rate of 575 babies per 10,000.



Figure 1

### NUMBER OF BABIES WITH BIRTH DEFECTS\* RHODE ISLAND, 1999-2003

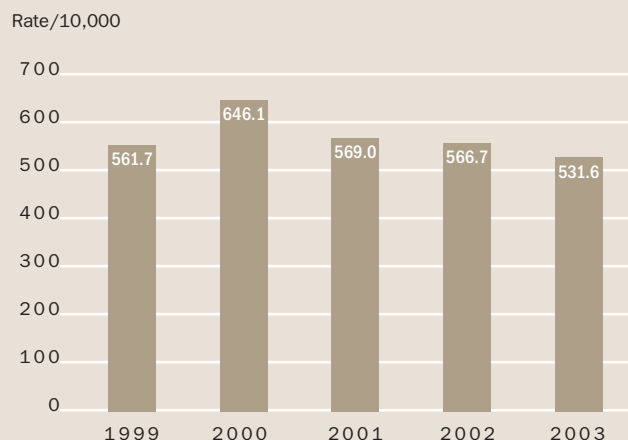


\*ICD-9-CM Codes: 740.0-759.9

Source: Hospital Discharge Database, Rhode Island Department of Health

Figure 2

### BIRTH DEFECT RATES RHODE ISLAND, 1999-2003



Source: Rhode Island Birth Defects Program

Division of Family Health, Rhode Island Department of Health

Tables 1 and 2 show the number and rate of selected birth defects among Rhode Island residents by organ system. The most frequent birth defects are those related to the cardiovascular system. The most common types of heart defects include: ventricular and atrial septal defects, endocardial cushion defect, and pulmonary valve atresia or stenosis. One out of every 39 babies born in Rhode Island is born with a defect related to the cardiovascular system.

Table 1

**BIRTH DEFECTS BY BODY SYSTEM, RHODE ISLAND, 1999-2003**

ORGAN SYSTEM	NUMBER	RATE PER 10,000
CARDIOVASCULAR	1562	254.3
MUSCULOSKELETAL/INTEGUMENTAL	1368	213.3
GENITOURINARY	805	138.2
GASTROINTESTINAL	232	36.1
EYE, EAR, FACE AND NECK	176	33.5
CENTRAL NERVOUS SYSTEM	105	18.7
CHROMOSOMAL	105	16.4
RESPIRATORY	72	12.0

*One out of every 39 babies  
born in Rhode Island is born with a defect  
related to the cardiovascular system.*



Table 2

**SELECTED BIRTH DEFECTS, RHODE ISLAND, 1999-2003**

BIRTH DEFECT	NUMBER	RATE PER 10,000 LIVE BIRTHS
<i>Central Nervous System</i>		
SPINA BIFIDA	20	3.3
ANENCEPHALY	2	0.3
ENCEPHALOCELE	2	0.3
<i>Eye</i>		
CONGENITAL CATARACT	7	1.1
ANOPHTHALMOS AND MICROPHTHALMUS	3	0.5
<i>Ear</i>		
MICROTIA	5	0.8
<i>Cardiovascular</i>		
TRANSPOSITION OF GREAT ARTERIES	11	1.8
TETRALOGY OF FALLOT	20	3.3
VENTRICULAR SEPTAL DEFECT	263	42.8
ATRIAL SEPTAL DEFECT	297	48.4
ENDOCARDIAL CUSHION DEFECT	7	1.1
PULMONARY VALVE ATRESIA/STENOSIS	79	12.9
TRICUSPID VALVE ATRESIA	6	1.0
AORTIC VALVE STENOSIS	10	1.6
HYPOPLASTIC LEFT HEART SYNDROME	9	1.5
COARCTATION OF AORTA	8	1.3
<i>Orofacial</i>		
CHOANAL ATRESIA	11	1.8
CLEFT LIP WITH AND WITHOUT PALATE	16	2.6
CLEFT PALATE	48	7.8
<i>Gastrointestinal</i>		
ESOPHAGEAL ATRESIA/TRACHEOSOPHAGEAL FISTULA	10	1.6
RECTAL AND LARGE INTESTINAL ATRESIA/STENOSIS	20	3.3
GASTROSCHISIS/OMPHALOCELE	33	5.4
<i>Genitourinary</i>		
HYPOSPADIAS AND EPISPADIAS	229	37.3
RENAL AGENESIS/HYPOPLASIA	15	2.4
OBSTRUCTIVE GENITOURINARY DEFECT	160	26.1
<i>Musculoskeletal</i>		
CLUB FOOT	133	21.7
REDUCTION DEFORMITY, UPPER LIMBS	11	1.8
REDUCTION DEFORMITY, LOWER LIMBS	4	0.7
DIAPHRAGMATIC HERNIA	8	1.3
<i>Chromosomal</i>		
DOWN SYNDROME	69	11.2
TRISOMY 13	5	0.8
TRISOMY 18	5	0.8
ALL BIRTH DEFECTS	3259	574.6

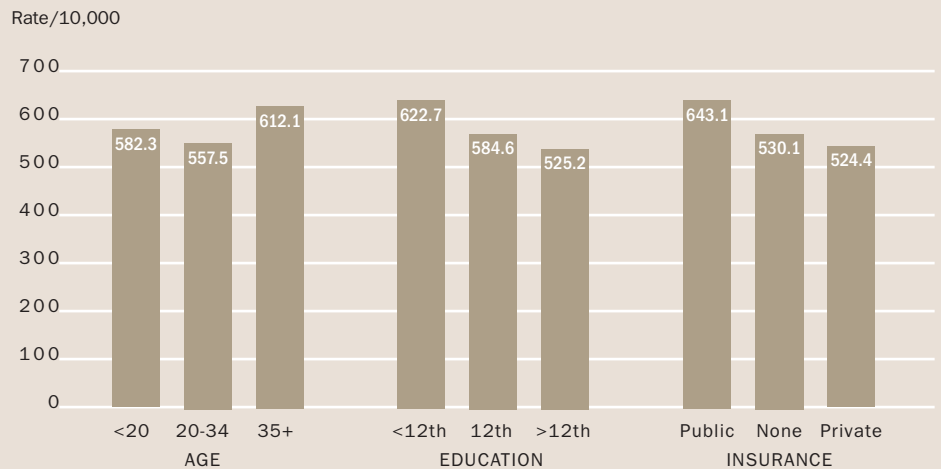


*Babies born to older women,  
women with less than a high school education, and  
women with publicly funded health insurance  
are at a higher risk for birth defects.*



Figure 3

**BIRTH DEFECT RATES BY SELECTED MATERNAL CHARACTERISTICS  
RHODE ISLAND, 1999-2003**



## MATERNAL CHARACTERISTICS

Babies born to older women (aged 35 or greater), women with less than a high school education, and women with publicly funded health insurance are at a higher risk for birth defects (Figure 3). During 1999-2003, the birth defect rate among women aged 35 or greater was 612.1 compared with 557.5 among women aged 20-34. Similarly, the birth defect rate among women with less than a high school education (622.7) was 1.2 times that of the rate among women with more than a high school education (525.2). Women who were insured through public programs, such as RItE Care and Medicaid were 1.2 times more likely to have a baby with a birth defect than women who were insured by commercial or private providers, such as Blue Cross and United HealthCare. The birth defect rate among babies born to women insured publicly was 643.1 compared with a rate of 524.4 among women with private insurance.





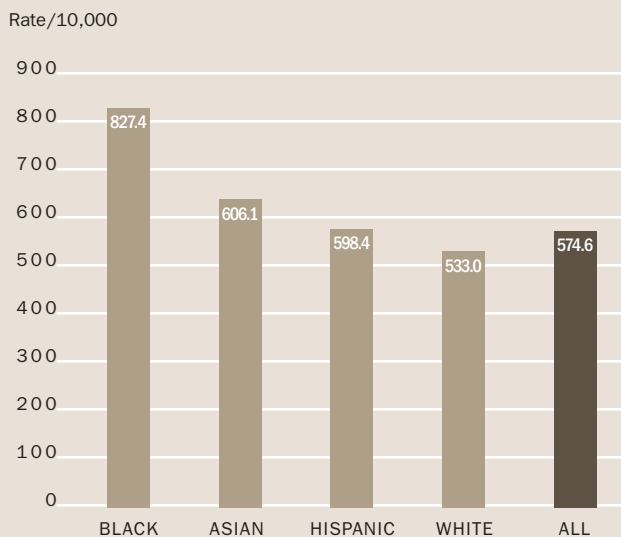
## RACIAL/ETHNIC AND GEOGRAPHIC DISPARITIES

Birth defect rates vary by race/ethnicity and geographical area. During 1999-2003, the average birth defect rate among African Americans was 827.4, 1.6 times the rate for Whites (533.0). Asians and those of Hispanic/Latino ethnicity also had birth defect rates that were higher than the rate for Whites (606.1 and 598.4, respectively). Figure 4 compares birth defect rates by race/ethnicity and geographical areas.

Birth defect rates were also higher among those who reside in Rhode Island's six core cities (Central Falls, Newport, Pawtucket, Providence, West Warwick, and Woonsocket) than those who live in the rest of the state (Figure 5). Babies born to residents of the core cities were 1.2 times more likely to have a birth defect than those born in the rest of the state (630.2 versus 530.8). Specifically, babies born to Newport residents had the highest birth defect rate, 888.9.

Figure 4

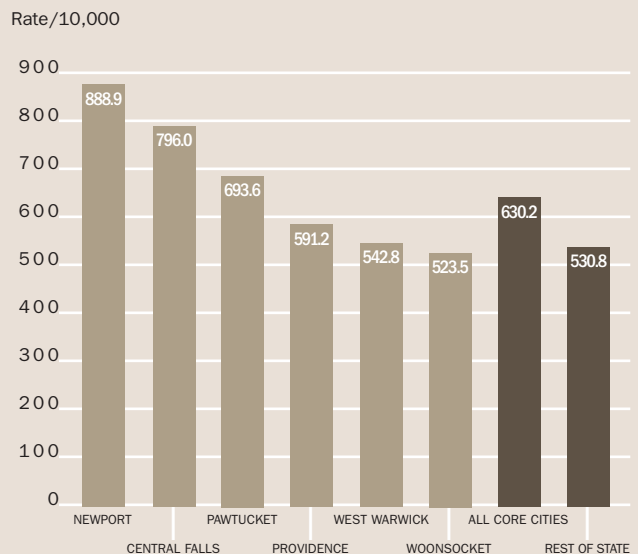
### BIRTH DEFECTS RATES BY RACE/ETHNICITY RHODE ISLAND, 1999-2003



Source: Rhode Island Birth Defects Program  
Division of Family Health, Rhode Island Department of Health

Figure 5

### BIRTH DEFECTS RATES BY SELECTED GEOGRAPHICAL AREAS, RHODE ISLAND, 1999-2003



Source: Rhode Island Birth Defects Program  
Division of Family Health, Rhode Island Department of Health

Table 3

**RHODE ISLAND BIRTH DEFECTS PROGRAM  
SENTINEL CONDITIONS LIST**

CONDITION	ICD-9-CM CODE
NEURAL TUBE DEFECTS	
» ANENCEPHALY	740.0-740.1
» SPINA BIFIDA	741.0-741.9
EYE	
» ANOPHTHALMIA/MICROPHTHALMIA	743.0, 743.1
» CONGENITAL CATARACT	743.30-743.34
» ANIRIDIA	743.45
CONGENITAL HEART DISEASE	745-747
CLEFT LIP/PALATE	749
ESOPHAGEAL ATRESIA/STENOSIS	750.3
ATRESIA/STENOSIS OF LARGE INTESTINE, RECTUM AND ANAL CANAL	751.2
GENITAL ANOMALIES	752
RENAL ANOMALIES	753.0-753.1
REDUCTION DEFORMITIES OF LIMBS	755.20-755.39
CONGENITAL DIAPHRAGMATIC HERNIA	756.6
GASTROSCHISIS/OMPHALOCELE	756.79
CHROMOSOMAL ANOMALIES	758
EAR	
» ANOTIA/MICROTIA	744.01, 744.23
» HEARING LOSS (REFERRAL VIA RI HEARING ASSESSMENT PROGRAM)	
LUNG	
» CONGENITAL CYSTIC LUNG	748.4
» AGENESIS, HYPOPLASIA, AND DYSPLASIA OF LUNG	748.5
CONGENITAL TUMORS	140-239
DEVELOPMENTAL CONDITIONS	
» AUTISM	299.0



## SENTINEL CONDITIONS

The Rhode Island Birth Defects Advisory Council identified a set of conditions for targeted outreach and follow-up to help assure appropriate services and referrals are provided. The selection of conditions was based on criteria that included the following: number of children affected; timeliness of identification; severity; service availability; resource intensity; recurrence risk; and availability of prevention strategies. The purpose of creating this list is to assure that families of children with these conditions are provided information and resources.



## COLLABORATIVE RESEARCH

### *Neural Tube Defects Study*

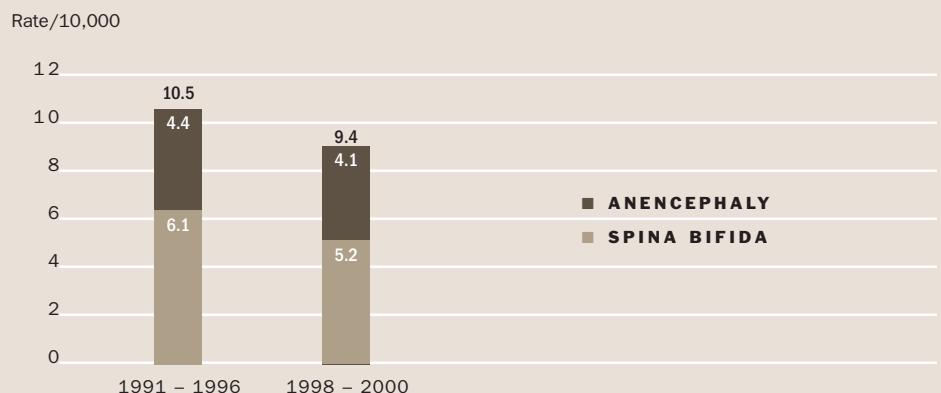
Studies have shown that folic acid supplementation can reduce the occurrence of neural tube defects (open spina bifida and anencephaly). A neural tube defects study was conducted in Rhode Island to determine the prevalence of neural tube defects, the impact of screening on prevalence, and the impact of folic acid fortification, which was introduced in 1997. The project was coordinated by the Foundation for Blood Research in Maine and was a collaboration between Maine researchers, Women and Infants Hospital, Rhode Island Hospital, and the Rhode Island Department of Health. Screening, termination (induced abortion), and live birth data were compared for two periods: 1991-1996 and 1998-2000. Data show that the rate of neural tube defects in Rhode Island decreased by 10.5%, from 10.5/10,000 before folic acid fortification (1991-1996) to 9.4 after folic acid fortification (1998-2000). Specifically, the prevalence of spina bifida decreased by 14.8%, from 6.1 to 5.2, and the prevalence of anencephaly decreased by 6.8%, from 4.4 to 4.1.



*Studies have shown that folic acid supplementation can reduce the occurrence of neural tube defects.*

Figure 6

#### **PREVALENCE OF OPEN NEURAL TUBE DEFECTS PRE AND POST FOLIC ACID FORTIFICATION RHODE ISLAND, 1991-1996 AND 1998-2000**



Source: Women and Infants Hospital, Rhode Island Hospital, Rhode Island Department of Health and Maine Foundation for Blood Research

*National and International Studies*

The Rhode Island Birth Defects Program is participating in a variety of national and international collaborative studies to gain a better understanding of specific birth defects, their trends, and associated factors. Examples of these collaborative research studies, their organizational sponsor, and purpose are described in Table 4.

Table 4

**COLLABORATIVE RESEARCH STUDIES**

STUDY/PROJECT	SPONSOR	PURPOSE/RESEARCH QUESTIONS
International Database on Craniofacial Anomalies (IDCFA)	World Health Organization (WHO)	To collect and disseminate data on craniofacial anomalies (oral clefts) to stimulate research for the development of prevention strategies and a better understanding of the characteristics associated with these anomalies.
Preterm Births and Birth Defects	National Birth Defects Prevention Network (NBDPN)	To determine what percentage of preterm and low birth weight births have birth defects; and whether there is an increased rate of preterm delivery for infants born with birth defects.
Neural Tube Defects and Infant Mortality	National Birth Defects Prevention Network (NBDPN)	To examine the impact of folic acid fortification on neural tube defect-specific infant mortality; whether a decline in neural tube defect-specific infant mortality has contributed to the decline in overall infant mortality; and if the severity of neural tube defects has changed over time.
Gastroschisis/Ventral Wall Defects	National Birth Defects Prevention Network (NBDPN)	To determine the prevalence and trends of specific ventral wall defects in the United States.
National Prevalence Estimates	National Birth Defects Prevention Network (NBDPN)	To monitor trends and determine differences between states and regions.

RESOURCES

Many children with birth defects are evaluated and followed at the Genetics Counseling and Child Development Centers at Rhode Island Hospital. Clinics are available for most birth defects, including meningomyelocele, Down Syndrome, inborn errors of metabolism, and other syndromes. The Cardiology Clinic is a pediatric specialty clinic at Hasbro Children’s Hospital. Rare conditions are referred to Boston specialists.

## ADDITIONAL INFORMATION: RELATED SOURCES AND WEBSITES

### **RHODE ISLAND ORGANIZATIONS:**

#### **Rhode Island March of Dimes**

<http://www.marchofdimes.com>

The mission of the March of Dimes is to improve the health of babies by preventing birth defects and infant mortality through research, community services, education, and advocacy.

#### **Rhode Island Parent Information Network (RIPIN) and Family Voices of Rhode Island**

<http://www.ripin.org>

Provides information, training, support, and advocacy to parents seeking help for their children.

### **NATIONAL ORGANIZATIONS:**

#### **National Birth Defects Prevention Network**

<http://www.nbdpn.org>

A national network of state and population-based programs for birth defects surveillance and research to assess the impact of birth defects upon children, families, and health care; to identify factors that can be used to develop primary prevention strategies; and to assist families and their providers in secondary disabilities prevention.

#### **National Center on Birth Defects and Developmental Disabilities**

<http://www.cdc.gov/ncbddd/>

Provides general information on birth defects, surveillance, and key research findings and risk factors.

#### **International Clearinghouse for Birth Defects Monitoring Systems**

<http://www.icbd.org>

Dedicated to the sharing of data, news, and views on congenital malformations monitoring, research, and prevention.

#### **National Down Syndrome Society**

<http://www.ndss.org>

Presents comprehensive information on Down Syndrome for families, health care professionals, educators, scientists, affiliates, parent support groups, and other organizations.

#### **Organization for Teratology Information Services (OTIS)**

<http://www.otispregnancy.org>

Studies the effects that drugs, medications, chemicals, and other exposures may have on the fetus. Provides resources for medical consultation on prenatal exposures and fact sheets about exposures that are known to cause birth defects.

### **SMILES**

<http://www.cleft.org>

SMILES is a group of dedicated families who have developed a first-hand understanding of the needs of children with cleft lip, cleft palate, and craniofacial deformities.

#### **Spina Bifida Association of America**

<http://www.sbaa.org>

Promotes the prevention of spina bifida and enhancing the lives of all affected.

#### **Teratology Society**

<http://www.teratology.org>

A scientific organization to study the causes and biological processes leading to abnormal development and birth defects.